SLC5A1 gene

solute carrier family 5 member 1

Normal Function

The *SLC5A1* gene provides instructions for producing a sodium/glucose cotransporter protein called SGLT1. This protein is found mainly in the intestinal tract and, to a lesser extent, in the kidneys, where it is involved in transporting glucose and the structurally similar galactose across cell membranes. Glucose and galactose are called simple sugars, or monosaccharides. They are obtained directly from the diet and from the breakdown of larger sugars and carbohydrates.

The sodium/glucose cotransporter protein is important in the functioning of the intestinal epithelial cells, which are cells that line the walls of the intestine. These cells have fingerlike projections called microvilli that absorb nutrients from food as it passes through the intestine. Based on their appearance, groups of these microvilli are known collectively as the brush border. The sodium/glucose cotransporter protein is involved in the process of using energy to move glucose and galactose across the brush border membrane for absorption, a mechanism called active transport. Sodium and water are transported across the brush border along with the sugars in this process.

Health Conditions Related to Genetic Changes

glucose-galactose malabsorption

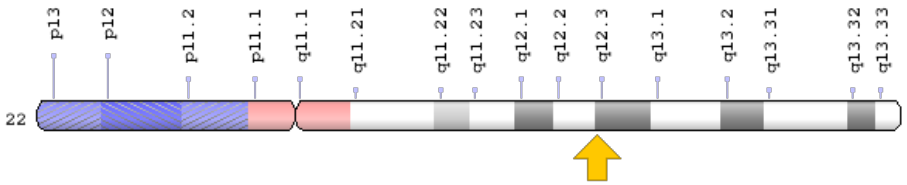
More than 40 mutations that cause glucose-galactose malabsorption have been identified in the *SLC5A1* gene. Some mutations result in a protein that is too short and does not function. Other mutations result in a protein that is of normal length but that is not folded properly, preventing it from being moved to the cell membrane where it is needed.

Mutations that prevent the sodium/glucose cotransporter protein from performing its function result in a buildup of glucose and galactose in the intestinal tract. This failure of active transport prevents the glucose and galactose from being absorbed and providing nourishment to the body. In addition, the water that normally would have been transported across the brush border with the sugar instead remains in the intestinal tract to be excreted with the stool, resulting in dehydration of the body's tissues and severe diarrhea.

Chromosomal Location

Cytogenetic Location: 22q12.3, which is the long (q) arm of chromosome 22 at position 12.3

Molecular Location: base pairs 32,043,032 to 32,113,029 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- D22S675
- Human Na+/glucose cotransporter 1 mRNA, complete cds
- NAGT
- SC5A1_HUMAN
- SGLT1
- solute carrier family 5 (sodium/glucose cotransporter), member 1
- solute carrier family 5 (sodium/glucose transporter), member 1

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC5A1%5BTIAB%5D%29+OR+%28%28NAGT%5BTIAB%5D%29+OR+%28SGLT1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SOLUTE CARRIER FAMILY 5 (SODIUM/GLUCOSE COTRANSPORTER), MEMBER 1
  http://omim.org/entry/182380

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SLC5A1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC5A1%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11036
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6523
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